



NAGA gene

alpha-N-acetylgalactosaminidase

Normal Function

The *NAGA* gene provides instructions for making the enzyme alpha-N-acetylgalactosaminidase. This enzyme works in the lysosomes, which are compartments within cells that digest and recycle materials. Within lysosomes, the enzyme helps break down complexes called glycoproteins and glycolipids, which consist of sugar molecules attached to certain proteins and fats. Specifically, alpha-N-acetylgalactosaminidase helps remove a molecule called alpha-N-acetylgalactosamine from sugars in these complexes.

Health Conditions Related to Genetic Changes

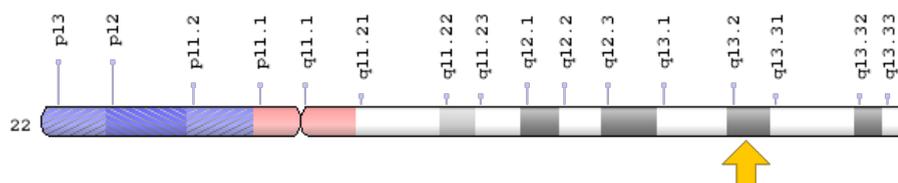
Schindler disease

Approximately seven *NAGA* gene mutations have been identified in people with Schindler disease. Most of these mutations are believed to change the 3-dimensional shape of the alpha-N-acetylgalactosaminidase enzyme, interfering with its ability to break down glycoproteins and glycolipids. These substances accumulate in the lysosomes and cause cells to malfunction and eventually die. Cell damage in the various tissues and organs of the body leads to neurological problems and the other signs and symptoms of Schindler disease.

Chromosomal Location

Cytogenetic Location: 22q13.2, which is the long (q) arm of chromosome 22 at position 13.2

Molecular Location: base pairs 42,058,334 to 42,070,870 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Acetylgalactosaminidase, alpha-N- (alpha-galactosidase B)
- alpha-N-acetylgalactosaminidase precursor
- D22S674
- GALB
- N-acetylgalactosaminidase, alpha-
- NAGAB_HUMAN

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Degradation of O-Glycans
<https://www.ncbi.nlm.nih.gov/books/NBK1934/#ch41.s6>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NAGA%5BTIAB%5D%29+OR+%28%28alpha-N-acetylgalactosaminidase%5BTIAB%5D%29+OR+%28alpha-N-acetylgalactosaminidase+precursor%5BTIAB%5D%29+OR+%28GALB%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

OMIM

- N-ACETYL-ALPHA-D-GALACTOSAMINIDASE
<http://omim.org/entry/104170>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NAGA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NAGA%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7631
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4668>
- UniProt
<http://www.uniprot.org/uniprot/P17050>

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